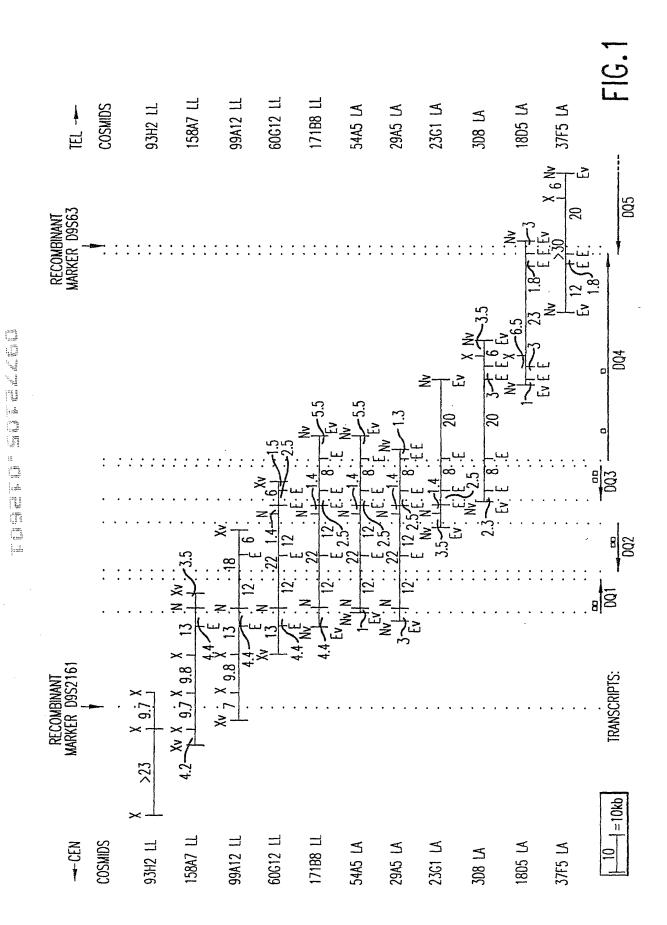
"Torsin, Torsin-Related Genes and Methods..."

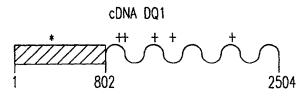
Inventors: Laurie J. Ozelius et al.



Docket No.: 0838.1001-009

"Torsin, Torsin-Related Genes and Methods..."

Inventors: Laurie J. Ozelius et al.

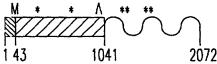


Polymorphisms: C/T @ 343, proline/proline

Transcript size: 2.7kb

Transcript size: 2.2kb, 1.4kb





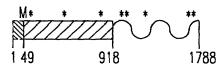
Polymorphisms: C/T @ 288, alanine/alanine

G/C @ 688, aspartic acid/histidine

G/T @ 1232 C/G @ 1255 del/T @ 1464 T/A @ 1495

Mutation: del/GAG @ 946, del/glutamic acid

cDNA DQ3



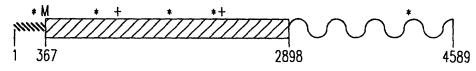
Polymorphisms: A/G @ 156, glutamic acid/glutamic acid

A/G @ 420, lysine/lysine T/C @ 801, glycine/glycine

AC/CT @ 1005 G/A @ 1063 (Ť)n @ 1273 T/A @ 1724

A/G @ 1751

cDNA DQ4



Polymorphisms: G/A @ 225

C/T @ 840, alanine/alanine G/A @ 1696, valine/isoleucine C/T @ 2172, histidine/histidine G/A @ 4225

Transcript size: 4.5kb

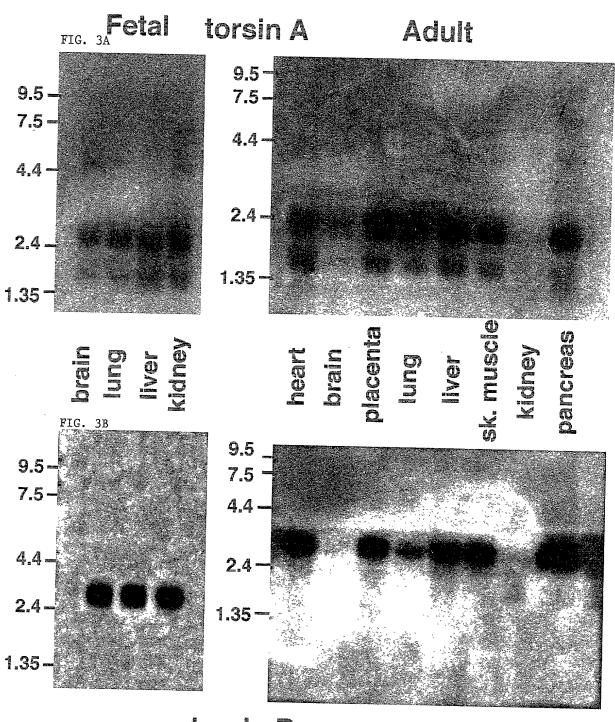
Transcript size: 1.8kb

FIG.2

0838.1001-009

"Torsin, Torsin-Related Genes and Methods..."

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torsin B

** *	Torging	THE CALL TO THE CALL	HICF VNTEL I SVITGK I KDSGTTI A I SAGAF WGI K NPI KCYI VECCUCDDAMICI W. 15: 5: 5: 5: 5: 5: 5: 5: 5: 5: 5: 5: 5	I OFFI	LECDL AGHL AKAL VVKSL KAFVQDP
MKI GRAVI GLI LI ADSVVVOA		MIMIZ DV VI	MINIMAR DI - VLLLLI HLCF VNTE		
TorsinA	Torsing	10000	ייין אינו	orpi	lorpe

KPKKPL TLSLHGVTGTGKNFVSKI I AENI YEGGLNSDYVHLFVATLHFPHASNI TL YKDQLQLVIRGNVSACARS IF IFDEMDKMHAGL I DA IKPFLD-YY NPKKPL TL SLHGVAGTGKÅNF VSQ I VAENLHPKGLKSNFVHLFVSTLHFPHEQK I KLYQDQLQKV I RGNVSACANSVFTFDEMDKLHPGI I DA I KPFLD-YY NPRKPLVLSFHGYTGSGKNYVAEIIANNTFRLGLRSTFVQHIVATNDFPDKNKLEEYQVELRNRILTTVQKCQRSIFIFDEADKLPEQLLGAIKPFLD-YY APSKPLVLSLHGVTGTGKSYVSSLLAQHLFRDGLRSPHVHHFSP11HFPHPSRTEQYKKELKSVVQGNLTACERSLFLFDEMDKLPPGLMEVLQPFLG-PS ----AAALHQTLF IFDEAEKLHPGLLEVLGPHLERRA ATP-BINDING DOMAIN **TorsinA [orsin**B TorpCel Torp1 **Torp**2

Docket No.:

Inventors:

0838.1001-009 "Torsin, Torsin-Related Genes and Methods

Laurie J. Ozelius et al.

ST I SGV DFRRS IF ILL SNKGGGE I AR I TKE QYESGYPREQL RLEAFEREL MNF SYNEK-GGL QMSEL I SNHL I DHF VPFL PL QREHVRSCVGAYL RKRGRG WVVYGTNYRKAIFIFISNAGGEQINQVALEAWRTNRDREEISLQEVEPVISRAVMDNPQHGFWRSGIMEEHLLDAVVPFLPLQRHHVRHCVLNELAQLGL-PEXXGLSL XWT IFLFL SNLRGD I INEVVLKLLKAGWSREE I TMEHLEPHLQAE I VDDHRQVL WHSRL VKENL I DYF I PFL PLEYRHVRLCARDAFL SQEL – EQVDGVSYXKAIFIFLSNAGGDLITKTALDFWRAGRKREDIQLKDLEPVLSVGVFNNKHSGLWHSGLIDKNLIDYFIPFLPLEYRHVKMCVRAEMRARGS DL VDGVSYQKAMFIFL SNAGAERIT DVAL DFWRSGKQREDIKLKDIEHAL SVSYFNNKNSGFWHSSLIDPNLIDYFVPFLPLEYKHLKMCIRVEMQSRGY orsinB TorpCel **TorsinA** [orp] **Forp**2

--LYKEETLDEIAQMMVYVPKEEQLFSSQGCKSIXQRIKLLPVMXG --EIDEDIVSRVAEEMTFFPKEERVF\SDK\GCKTVF\TKLD\YYYDD --AIDEDIVTRVAEEMTFFPRDEKIYSDKGCKTVQSRLDFH --EPARRWFRRCVTD-TYFPEVEQLFSSNGCKTVASRLTFF! DL VSNV DF VERVLNSL QYFPESSKAF SSSGCKRV TorsinB orsinA TorpCe (orp2 [orp]

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